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# PREVALENCE OF GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY IN A TERTIARY CARE HOSPITAL OF HIMALAYAN REGION.

Paediatrics

**Dr Chiranth R*** Senior Resident, Department of Pediatrics, BGS Global Institute Of Medical Sciences, Bengaluru . *Corresponding Author

**Dr Shayam Kaushik** Professor AND Head .Department of Pediatrics, Dr Y S Parmar Medical college , Nahan.

**Dr. Sunita** Professor, Department Of Biochemistry, IGMC, Shimla.

## ABSTRACT

**Background:** G6PD deficiency is most prevalent enzyme deficiency with an estimate of 400 million people all over the world. It is an inherited deficiency that is one of the cause of neonatal hyperbilirubinemia. As there were no studies conducted in the past regarding prevalence of G6PD deficiency in region of Shimla, Himachal Pradesh .The present study was conducted to determine the prevalence of G6PD deficiency in region of Shimla Himachal Pradesh, India.

**Methods:** Objective of the study:To find the prevalence of G6PD deficiency in hospital born babies and to assess its contribution in causing neonatal jaundice.Study site: Kamala Nehru State Hospital for Mother & Child. (KNSHM&C), IGMC, Shimla

**Study Design:** Prospective hospital based study.Study period: July 2017 to August 2018.

**Source Of Data:** Babies delivered in Kamala Nehru State Hospital for Mother & Child. (KNSHM&C), IGMC, Shimla

**Results:** The screening for g6PD deficiency was done by WST-8 Formazan method.The prevalence of G6PD deficiency was found to be 0.5%

**Conclusion:** Results of the study indicate that prevalence of G6PD deficiency in Shimla region of Himachal Pradesh is 0.5% .However large scale population based studies must be taken into consideration for future planning of strategies for neonatal screening. The screening program could be introduced in all institutional deliveries at tertiary hospitals and then gradually scaled up to cover institutional deliveries over the entire country

## KEYWORDS

G6PD Deficiency, Screening Programme, Neonatal Jaundice.

## INTRODUCTION

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is an X linked inherited disorder commonly presenting as neonatal jaundice[1]. It is an enzyme in the pentose monophosphate pathway which catalyses the conversion of nicotinamide adenine dinucleotide phosphate (NADP) to its reduced form, NADPH. It protects the red blood cells (RBCs) from oxidative damage[2] . G6PD deficiencyis commonly seen in African, South East Asian and middle eastern populations[3,4]. In India, the incidence of G6PD deficiency varies from 0 to 27%[5] . There are 13 biochemical variants of G6PD being reported so far out of which Mediterranean type is most common. Among the tribal, Orissa variant is frequently detected. The third common variant is Kerala Kalyan type[5] . G6PD deficiency is one of the important causes of severe neonatal jaundice, which can lead to kernicterus[6] . If detected early, children with this disorder can be reared with all the necessary precautions to lead a normal and healthy life. Hence, G6PD screening should be further reinforced for every newborn. The objective of this study was to find out the prevalence of G6PD deficiency in hospital born babies and to assess its contribution in causing neonatal jaundice.

## MATERIALS AND METHODS

This hospital based cross sectional study was conducted in Kamala Nehru State Hospital for Mother & Child. (KNSHM&C), IGMC, Shimla from July 2017 to August 2018 for one year. All newborns delivered at KNSHM&C Shimla formed the study cohort. Written informed consent for participation in the study was taken from either of the parents. Data regarding maternal age, consanguinity, gestation in weeks, gravid status, maternal blood group, drug intake during pregnancy, history of neonatal jaundice in previous sibling, history of early neonatal death was recorded in a structured case record proforma.

History regarding course of labour, fetal distress, type of delivery, maturity, Apgar scores at 1st and 5th min, requirement of resuscitation, vitamin k injection after birth was also be recorded. Blood samples were collected in EDTA vial from the umbilical cord immediately after the delivery. Cord blood was examined for G6PD deficiency using formazan method ( WST –Formazan method) [7] with in 48 hours.

### Principle Of The WST-8 Formazan Method And Preparation Of Reaction Mixtures

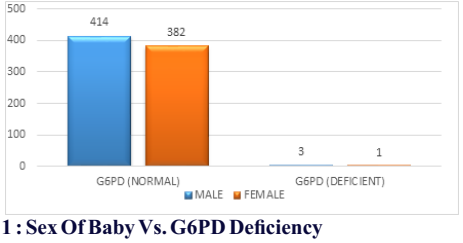
Briefly, the hydrogen of NADPH produced by G6PD converts WST-8 to WST-8 formazan in the presence of a hydrogen carrier, 1-methoxy PMS. The reaction mixtures included are: (1) 0.05 m Tris- HCl buffer (or 0.05 m HEPES buffer), adjusted pH to 7.2–7.5, which contained 5 mm MgCl2 and 0.1% saponin, (2) the substrate mixtures of 2.5 mm G6P, 0.2 mm NADP in H2O, and (3) the WST-8/1-methoxy PMS mixture. The WST-8/1-methoxy PMS mixture can be stored for 3 months at 4 deg C in the dark or for 12 months at -20 deg C, and other reagents for 6–12 months at 4 deg.C in the dark. All the reactions were carried out as Standard method in a micro tube or in a well plate as economic method along with controls, at room temperature. Five microlitres of blood were mixed with the reaction mixtures, and color photographs were taken at various intervals. These values were compared with those G-6-P controls. Those neonates who will be identified as positive for G6PD deficiency on the screening test observed up to one week of age for the appearance of hyperbilirubinemia and discharged after explaining the underlying disease and precautions (including the list of medicines ) to be taken to prevent haemolysis.

## RESULTS

In this study 800 newborns delivered in KNSHM&C Shimla were screened for G6PD deficiency from July 2017 to August 2018.. Four ( 0.5%) newborns out of 800 were found to have deficient G6PD activity.Of these 418 (52.2%) were males and 382 (47.8%) females. out of which 0.7% (3 males) and 0.2% ( 1 female) were found to be G6PD deficient .The difference was not statistically significant (P value of 0.626.)

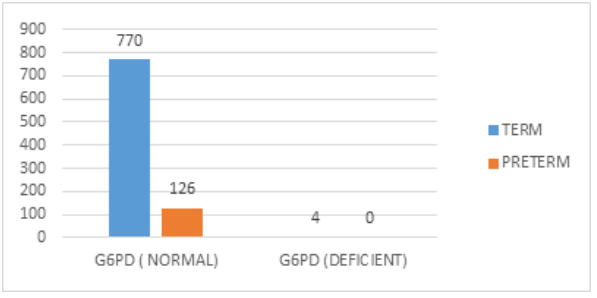
**Table 1 : Prevalence Of G6PD Deficiency In Neonates Delivered In KNH & SH, Shimla**

| No of babies screened | Method | No of babies with G6PD deficiency | Prevalence |
|---|---|---|---|
| 800 neonates | WST-8 Formazan method | 4 | 0.5 |



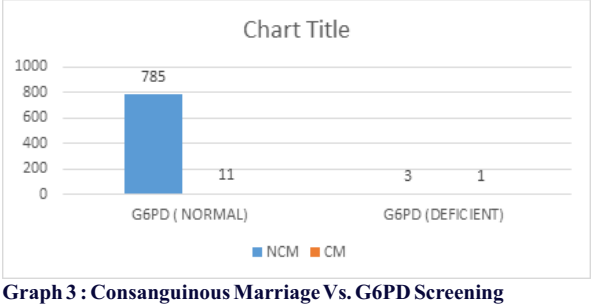

**Graph 1 : Sex Of Baby Vs. G6PD Deficiency**

In the present study out of 800 newborns screened, 674 were term (84.3%) and 126 (15.7%) preterm. Four term neonates were found to be G6PD deficient. All the preterm newborns screened had normal G6PD activity. Statistically no significance was found between gestational age and G6PD deficiency.



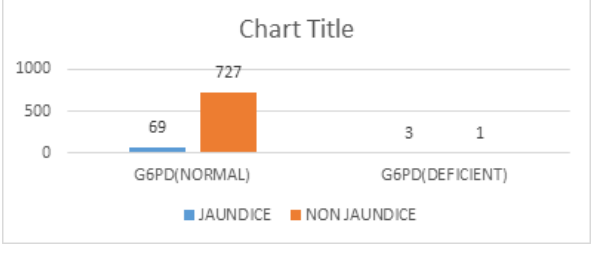

**Graph 2: Gestational Period Vs. G6PD Screening**

Among the screened , 788 ( 98.5%) were born to non consanguinous marriage and 12 (1.5%) born to consanguinous marrige. Three newborns (0.3%) born to non consanguinous marrige and one newborn (8.3%) born to consanguinous marriage were G6PD deficient. The difference was statistically significant (P value = 0.005).



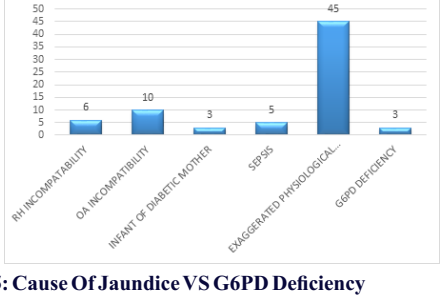

**Graph 3 : Consanguinous Marriage Vs. G6PD Screening**

Of all the newborns screened, four newborns were G6PD deficient and 796 newborns had normal G6PD activity .69 newborns (8.7%) newborns out of 796 who were G6PD normal developed pathological jaundice where as 3 out of 4 (75%) G6PD deficient newborns developed pathological jaundice. The prevalence of pathological jaundice was significantly higher in G6PD deficient newborns as compared to G6PD normal newborns ( P value = 0.003.)




**Graph 4 : Jaundice Vs. G6PD Screening**

Among the 72 newborns who developed pathological jaundice ,the most common cause of pathological jaundice was exaggerated physiological jaundice (62.5%) followed by OA incompatibility (13.8%) . Other causes of pathological jaundice were RH incompatibility (8.3%) , sepsis (6.9%) , G6PD deficiency (4.1%) and infant of diabetic mother (4.1%) .



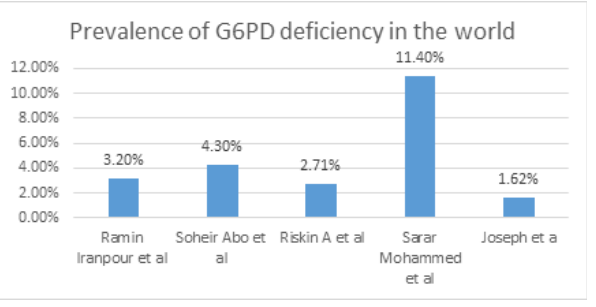

**Graph 5: Cause Of Jaundice VS G6PD Deficiency**

Out of 4 G6PD deficient neonates, 3 neonates developed pathological jaundice .Of these 2 neonates required only phototherapy and one neonate required exchange transfusion and phototherapy.

## DISCUSSION

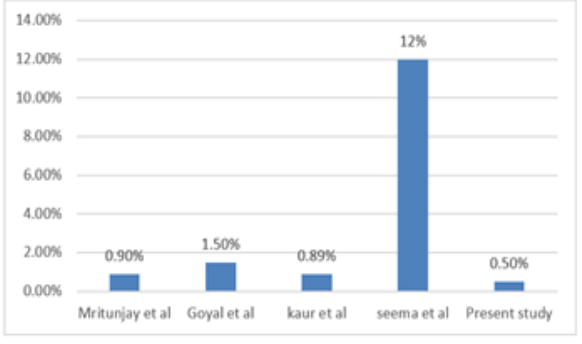
G6PD deficiency is a major risk factor for the development of neonatal jaundice and increases the risk for bilirubin neurotoxicity. The prevalence of G6PD deficiency is variable in different countries and different ethnic groups within a country. In the present study, 800 newborns delivered in Kamala Nehru State Hospital of Mother and Child (KNSHM&C), Shimla were screened. Four newborns were found to have deficient G6PD activity.

Prevalence of G6PD deficiency was found to be 0.50%. prevalence of G6PD deficiency is different across various parts of the world. Ramin Iranpour et al[1] (Iran, 2006) screened 2501 babies using a semi-quantitative assay, out of these 79 neonates were found to have G6PD deficiency with the prevalence of 3.2 %, Soheir Abo et al[8] (Egypt, 2015) screened 2782 newborns in Egypt in the year 2015 with the prevalence of G6PD deficiency of 4.3%,Riskin A et al [9] (Israel, 2012) screened 2656 newborns with an overall prevalence of G6PD deficiency of 2.71%,Joseph et al[10](Singapore, 1999) screened newborns in Singapore with the prevalence of G6PD deficiency of 1.62% and Sarar Mohammed et al ( Saudi Arabia ) screened 1366 newborns with the prevalence of G6PD deficiency 11.4%.



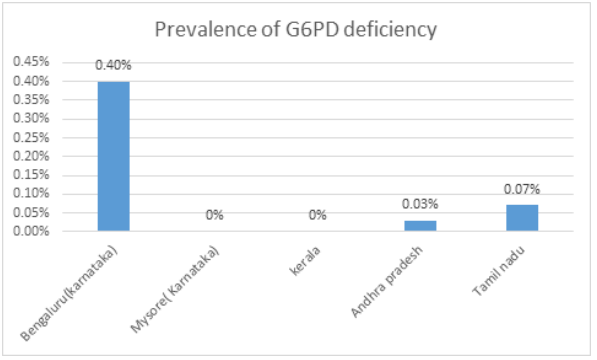

**Graph 6: Prevalence Of G6PD Deficiency By Various Studies In The World**

Prevalence of G6PD deficiency also varies in different parts of India. A study by Vandana rai et al showed the frequency of G6PD deficiency among the Indian population as a whole range from 0 to 27 %.G6PD – deficient frequency is comparatively higher in North and West Indian zones. In the study done by Mritunjay et al in Delhi , 62 out of 2479 babies screened for G6PD deficiency by a semiquantitative assay, 50 neonates were found to be G6PD deficiency with the prevalence of 2%. In a similar study done by Goyal et al showed a 1.50 % prevalence of G6PD deficiency in Delhi. The study by Kaur et al showed 0.89% deficient G6PD activity in Chandigarh, Punjab. The study by Rosy Khandela et al showed 1.92 % prevalence of G6PD deficiency in Guwahati. In the study done by Seema et al in Tanda Himachal Pradesh in 2011, out of 5652 babies screened for G6PD deficiency, 703 neonates were found to be G6PD deficient by WST- 8 formazan method with the prevalence of 12%.The prevalence of G6PD deficiency in our study is 0.5% which is comparable to the study conducted by Mritunjay et al, Goyal et al, Kaur et al, and Rosy Khandela et al while low comparedto Seema et al in Tanda, Himachal Pradesh. This could be due to non-uniform sample size, geographical and racial variations.




**Graph 7 : Prevalence Of G6PD Deficiency By Various Studies In North India**

The prevalence of G6PD deficiency is low in south India when compared to North and West India. The study by M K Mohinuddin et al showed 0.4% deficient G6PD activity in Bengaluru, Karnataka. Similar studies by Chandrashekar et al in Mysore Karnataka and Hakim et al in Kerala showed 0% prevalence of G6PD deficiency. Regional distribution of G6PD deficiency studied by Bhasin et al[62] 0.03% in Andhra Pradesh and 0.07% in Tamilnadu.




**Graph 8 : Prevalence Of G6PD Deficiency By Various Studies In South India**

Among the newborns screened 418 ( 52.2% ) were males and 382 ( 47.8% ) females .out of which 0.7% (3 males) and 0.2% (1 female) were found to be G6PD deficient. All the other studies showed the prevalence of G6PD deficiency is higher in males compared to females.As G6PD deficiency is X-linked recessive disease, the prevalence of G6PD deficiency was significantly higher in males than in females. Males are affected by X-linked recessive disorders much more common than females. In males (who have only one X chromosome), one altered copy of the gene is sufficient to cause the disease. In females ( who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disease

The present study also collected the data of gestational age.out of 800 babies screened, 674 were term (84.3%) and 126 ( 15.7%) were preterm . Four term neonates were found to have deficient G6PD activity. Ramin Iranpour et al (Iran, 2006) et al screened 2501 babies out of which, 2249 (89.9%) were term and 202 (8.07%) were preterm babies.68 term babies (2.9%) and 11 preterm (5.4%) were found to be G6PD deficient. This is in concordance with our study as no statistical gestational age difference was found between term and preterm newborns.

Among the neonates screened, 12 (1.5%) were born to consanguineous marriage and 1 (8.3 %) were found to be G6PD deficient. Out of 788 (98.5%) babies born to non-consanguineous marriage, 3 babies (0.3%) were G6PD deficient.In the study by Sukumal et al , out of 109 babies screened, 5 (4.6%) were born to the consanginous married couple and 1 (20%) was found to be G6PD deficient. Out of 104 (95.4%) babies born from non-consanguineous marriage, 15 babies (14.42%) were G6PD deficient.

In the present study out of 800 newborns screened for G6PD deficiency, 9% ( 72 newborns) developed pathological jaundice. out of 4 G6PD deficient newborns, 3 ( 75%) developed pathological jaundice .% of G6PD normal newborns developed pathological jaundice was 8.6% ( 69 newborns).In the similar studies done by Mritunjay et al, Rosy kandela et al and Seema et al percentage of G6PD deficient newborns developed pathological jaundice were 32%, 41.6%, and 45% respectively.

## CONCLUSION

In this study, the applicability of a microtubes mass-screening assay to provide prevalence of G6PD deficiency in Himachal Pradesh was evaluated. This study showed that it is possible to undertake a large-scale mass screening of G6PD deficiency using the microtubes assay in order to assess prevalence of the deficiency. Results of the study indicate that prevalence of G6PD deficiency in Shimla region of Himachal Pradesh is 0.5% .However large scale population based studies must be taken into consideration for future planning of strategies for neonatal screening. The screening program could be introduced in all institutional deliveries at tertiary hospitals and then gradually scaled up to cover institutional deliveries over the entire country.

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